EXECUTIVE SUMMARY

FARMINDUSTRIA

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Rare Diseases: a socially important rarity

A rare disease is a disease with an incidence of fewer than **5 cases per 10,000** persons.
Rare diseases number between **5,000 and 8,000** worldwide and mainly affect **children** who account **for between 50% and 75%** of all patients. The rarity of such diseases is often a complicating factor in the research for pharmaceutical treatments.

Therefore, news of a **medicine that can finally treat a rare disease** is always greeted with **unforgettable emotion** among rare-disease patients who have had, often since birth, to coexist with the pathology. Such treatment, by removing the suffering and limits frequently determined by rare diseases, is a first step toward a better quality of life. And sometimes **"this" new treatment** can offer a **realistic hope** of recovery.

Access to therapy: a right not fully exercisable in many parts of the country

As rare-disease patients must have immediate access to rare-disease therapies that often constitute **the only available treatment**, such access represents a **non-deferable right**.

Therefore, whatever hinders the exercise of this right adversely affects both patients and their families.
But why are pharmacological therapies authorised nationally by the Italian Medicines Agency **not equally and/or promptly accessible** throughout the entire country?
Generally, the reason is to be found in the **purely economistic calculations** applied by regional, provincial and hospital-trust authorities that **limit** or **delay the use** of these medicines, especially as regards adults afflicted by rare diseases.

And this attitude persists despite the fact that a **National Rare Diseases Network** has been in place since 2001, comprising **all the facilities and services of the regional health systems** and set up to implement prevention and surveillance actions, improve diagnosis and treatment and promote information and training; in other words, **a network for the protection of rare disease patients**.
In many parts of the country far too **many differences** exist in the relations between healthcare centres and territorial assistance facilities that, together with other deficiencies, prevent rare-disease patients from enjoying equal access to the medicines available.

Similarly, a **National Rare Diseases Register** has been set up to collate data from the regional or interregional registers, but **it is still beset by serious drawbacks**, especially completeness and updating.

European regulations to safeguard research and rare-disease patients

In view of the small number of patients and limited experience available, the EU deemed that it was essential to set up a **European Rare Diseases Reference Network** to guarantee the diagnosis and treatment of these pathologies. At present **24 European reference networks** are operational, with which **Italy participates in 23**.

Regulation (EC no. 141/2000) by **recognizing the special nature of rare-diseases R&D** offers regulatory benefits to companies engaged in the research and development of orphan medicines for the precise purpose of improving rare-disease healthcare.

The commitment of Aifa, the Italian regions and pharmaceutical companies during the pandemic

Since the onset of the pandemic, **Aifa, the regions and pharmaceutical companies** have done their utmost to **accommodate** the fragility of rare-disease patients, and in this context several specific measures have been introduced.

1. **Pharmaceutical companies** have **enhanced patient support programmes and home therapy**, whose numerous advantages include facilitating therapeutic compliance and access to medicines, improving the clinical monitoring of the pathology and simplifying caregivers' everyday activities.

2. **The Italian Medicines Agency** has **extended the validity of its therapeutic plans** for medicinal products subject to monitoring and has widened access to home therapy, thus minimizing the potential risks deriving from discontinuities in treatment attributable to the fear of contracting contagion in a hospital environment.

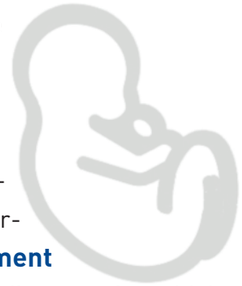
3. **Some regions** have implemented measures to expedite access to therapy by **shifting the distribution of A-PHT medicines from direct distribution to pharmacy-facilitated distribution**.

The provision of **care to rare-disease patients** also entails the provision of a **caregiver**, a figure whose services are of great social and economic value for the country.
Given their key role, **training** caregivers is increasingly **important** to improve their skills and knowledge in respect of the pathology, patient rights and the socio-health and welfare networks in whose framework they operate.

Early diagnosis: a fundamental tool

As we are dealing with complex and strongly debilitating pathologies, **early diagnosis** is **extremely important** in helping to reduce the symptoms and complications associated with the natural progression of the disease and/or patients' death.

Neonatal screening is therefore of fundamental importance for **the timely treatment** of patients suffering from diseases for which research has found effective therapies that can prevent degeneration.



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Research: patient scarcity and difficulties to be overcome jointly

Rare diseases are a **specialised research area for pharmaceutical companies**, and one that raises specific problems deriving from the limited numbers of people affected by each rare pathology, the paucity of specialised centres, the limited nature of the epidemiological data available and, not least, the difficulties in understanding the diseases themselves.
Precisely for this reason it is necessary to **apply research methodologies** that can enhance the **efficiency** of design studies and data analysis, **mitigate recruitment difficulties** and **minimize the rates** of logistically-related **patient dropouts** during the study.

Dialogue and **open cooperation between the various parties** involved in the macro-process of clinical research - regulatory authorities and the institutions, patient associations, scientific companies, researchers and pharmaceutical companies - are fundamental factors in **tackling the main challenges** posed by rare diseases in terms of managing clinical studies and **identifying more innovative approaches** for redesigning future clinical research.

Urgent actions to be taken in the interest of patients

The health emergency facing our country calls for a rapid overhaul of the all system if we are to improve the access to and availability of therapies and healthcare services for patients, and which would, of course, also entail extremely important innovations in the treatment of rare-disease patients.
Pharmaceutical companies wish to work **side-by-side with patients and the institutions** so that they may make their own contribution towards the definition of a forward-looking and integrated strategy for the complex world of rare diseases.

It is an encouraging **signal** that Parliament is now debating a **bill** designed to sustain research and production for orphan medicinal products as well as care for rare diseases (TU C. 164 and abb.) as it means that the institutions are now **paying attention** to the problem while also creating **an opportunity** for all sector participants.

Measures that can help to improve the lives of many rare-disease patients

In the light of the experience gained during this period, it would be highly desirable to quickly implement the following **priority measures** in order to **provide** stable **access to available therapy and care services** for rare-disease patients.

SHORT-TERM MEASURES

- Facilitating immediate access to available therapies for rare-disease patients by heightening the awareness of:

  a. the Social Affairs Committee of the Chamber of Deputies on the need to redraft article 5, subsection 3, of the bill designed to sustain research and production for orphan medicinal products as well as care for rare diseases by eliminating all references to law no. 648/96 and thus ensuring that all **medicinal products authorised** by AIFA for the treatment of rare diseases can be **made available** by the regions **regardless of whether or not they are included in hospital formularies** or similar lists;

  b. the Italian Medicines Agency on the importance of **complying with the 100-day deadline** for concluding reimbursability and pricing procedure as laid down for orphan medicinal products and medical products of exceptional therapeutic importance.

- Heightening the Health Ministry's and Parliament's awareness of the importance of a **clear regulatory framework** to allow recourse to **home therapy** throughout the entire national territory, and a framework, moreover, that the regions would have to adopt so as to guarantee equality in the treatment of rare-disease patients while also offering the possibility of **public-private partnerships**.

- Making the Health Ministry aware of the importance of fully implementing law no. 167/2006, by convening, **at the earliest possible date**, a **working table** tasked to review the panel of **pathologies subject to neonatal screening** (genetic neuromuscular diseases, severe congenital immunodeficiencies and lysosomal storage diseases) and put arrangements in place for the panel's **annual update**.

MEDIUM-TERM MEASURES

- Heightening the Health Ministry's awareness of the need to **approve the new Rare-Diseases National Plan**, in concert with the State-Region Conference, by providing adequate funding and initiating constructive dialogue with all stakeholders.

- Informing the Health Ministry committee tasked to update the L.E.As (basic healthcare levels) about the importance of **a procedure to facilitate the inclusion of rare diseases among such basic healthcare levels**.

- Reminding the Health Ministry of the need to **create** and **standardize therapeutic diagnostic assistance procedures** by laying down appropriate general guidelines, to be adapted and supplemented according to pathology, through the adoption of national regulations.

LONG-TERM MEASURES

- Emphasizing the importance of adopting more effective **monitoring** by **Regional and/or Interregional Coordination Centres** and, where necessary, providing support to healthcare centres and territorial facilities.

- Making the regions duly aware of the advantages of **automatically inserting A-PHT medicinal products** for rare diseases in the list of **medicines for pharmacy-facilitated distribution** in order to redistribute healthcare assistance across the territory and ensure the widespread distribution of these medicines in all regions.

- Publicising the fact that the **pharmaceutical companies are willing** to continue their **full collaboration** with the **regulatory authorities and institutions, patient associations, scientific companies and researchers** in order to address the main challenges posed by clinical trials for rare diseases in the best way possible and introduce more innovative approaches.

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